AMSER Rad Path
Case of the Month:

Painful Mass in 64 yo Male with Neurofibromatosis Type 1

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Patient Presentation

Clinical History:

- 64 year old male with a complex medical history including CAD, chronic bradycardia, hypertension, hyperlipidemia, asthma, OSA, hypothyroidism, osteoarthritis, and spongy lesions consistent with neurofibromas
- NF1 is associated with chronic pain. He has an implanted intrathecal pain pump which was repositioned 6 years ago
- He presented with a slowly enlarging mass in his abdominal wall where his intrathecal pump was previously inserted. This mass was painful and he requested removal.

Social History:

- 1/2 pack daily smoker with 10 year pack history
- No alcohol use

Physical Exam:

- Abdomen: soft, non-distended with a tender swollen subcutaneous lesion to R flank
- Skin: numerous diffuse soft spongy lesions consistent with history of NF1
- Remainder of exam was normal

What images should we order?

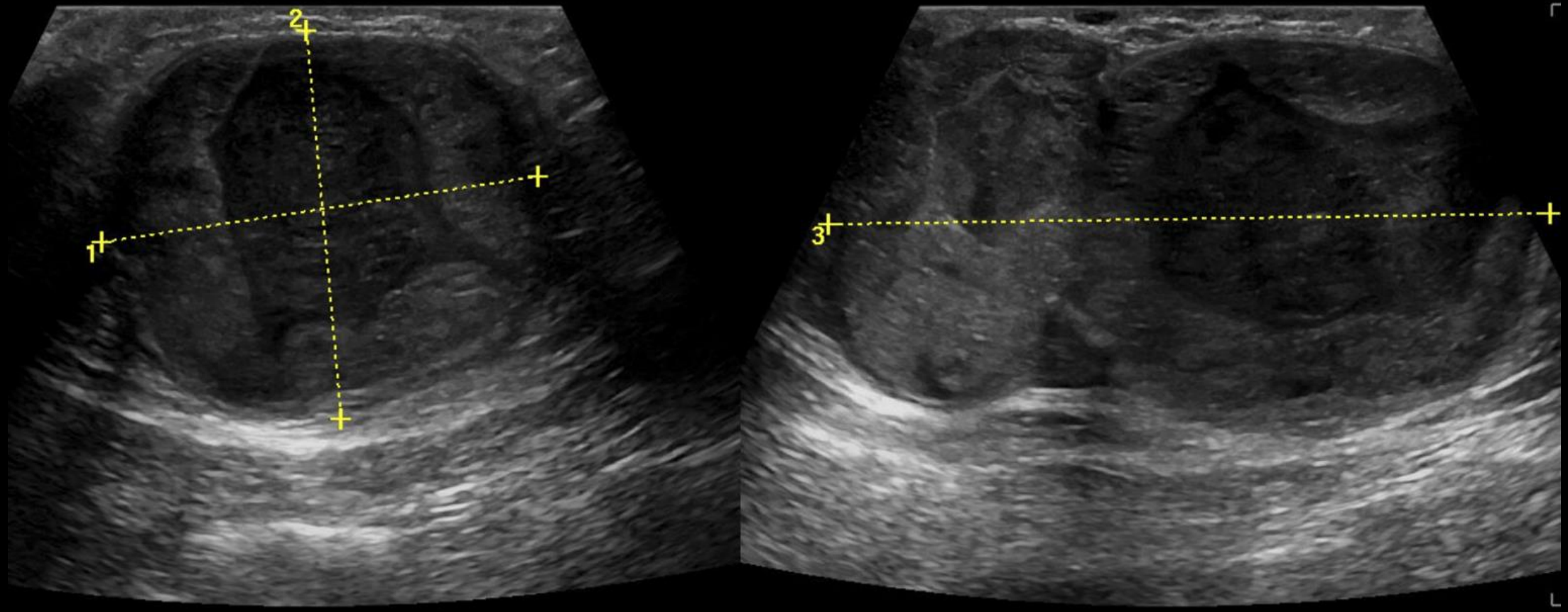
ACR Appropriateness

Variant 2: **Nonsuperficial (deep) soft tissue mass. Initial imaging.**

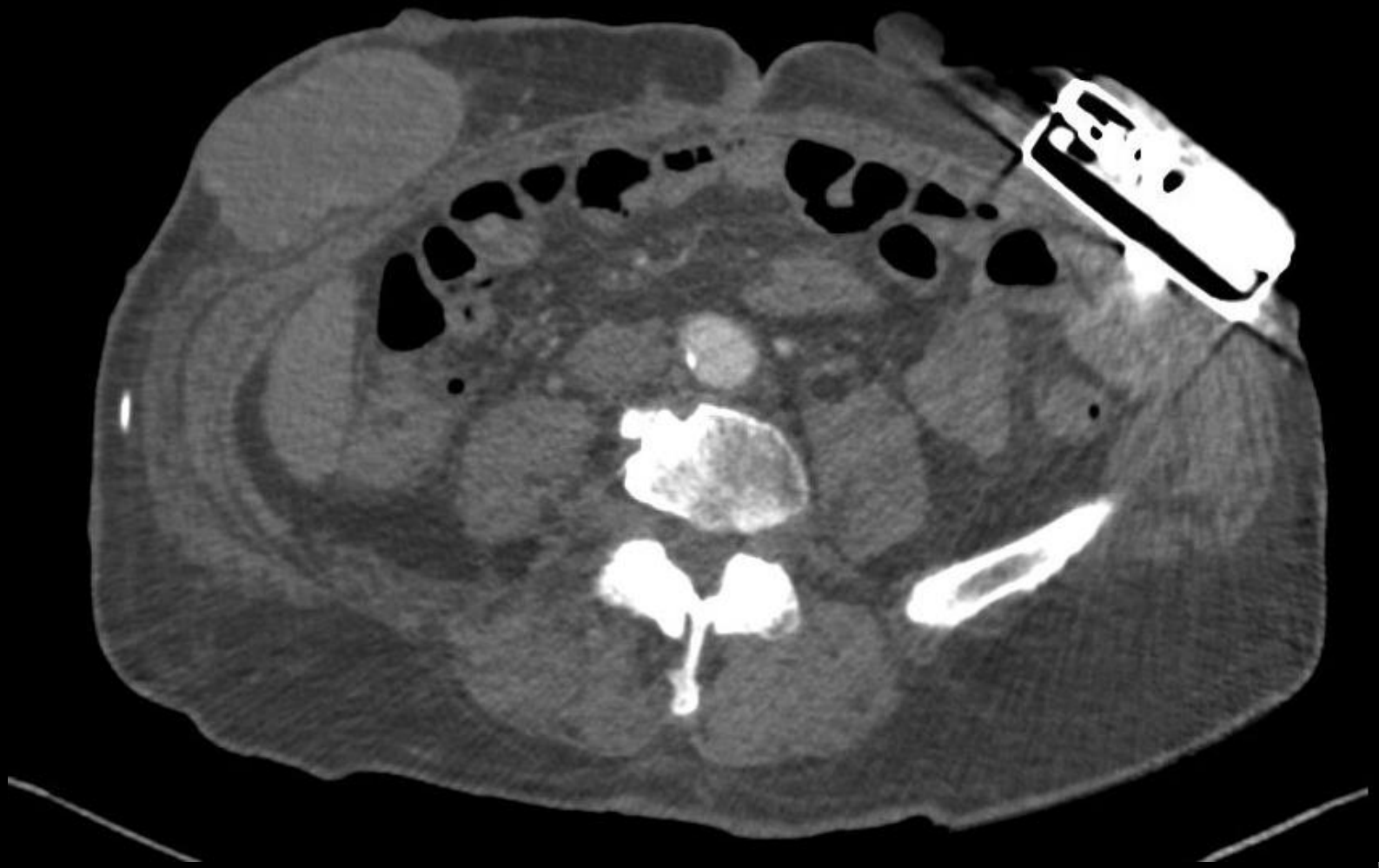
Procedure	Appropriateness Category	Relative Radiation Level
Radiography area of interest	Usually Appropriate	Varies
US area of interest	May Be Appropriate	O
CT area of interest with IV contrast	May Be Appropriate	Varies
CT area of interest without and with IV contrast	May Be Appropriate	Varies
CT area of interest without IV contrast	May Be Appropriate	Varies
US area of interest with IV contrast	Usually Not Appropriate	O
Image-guided biopsy area of interest	Usually Not Appropriate	Varies
Image-guided fine needle aspiration area of interest	Usually Not Appropriate	Varies
MRI area of interest without and with IV contrast	Usually Not Appropriate	O
MRI area of interest without IV contrast	Usually Not Appropriate	O
FDG-PET/CT area of interest	Usually Not Appropriate	☢☢☢☢

Per criteria, obtained a CT with/without IV contrast & US abdomen

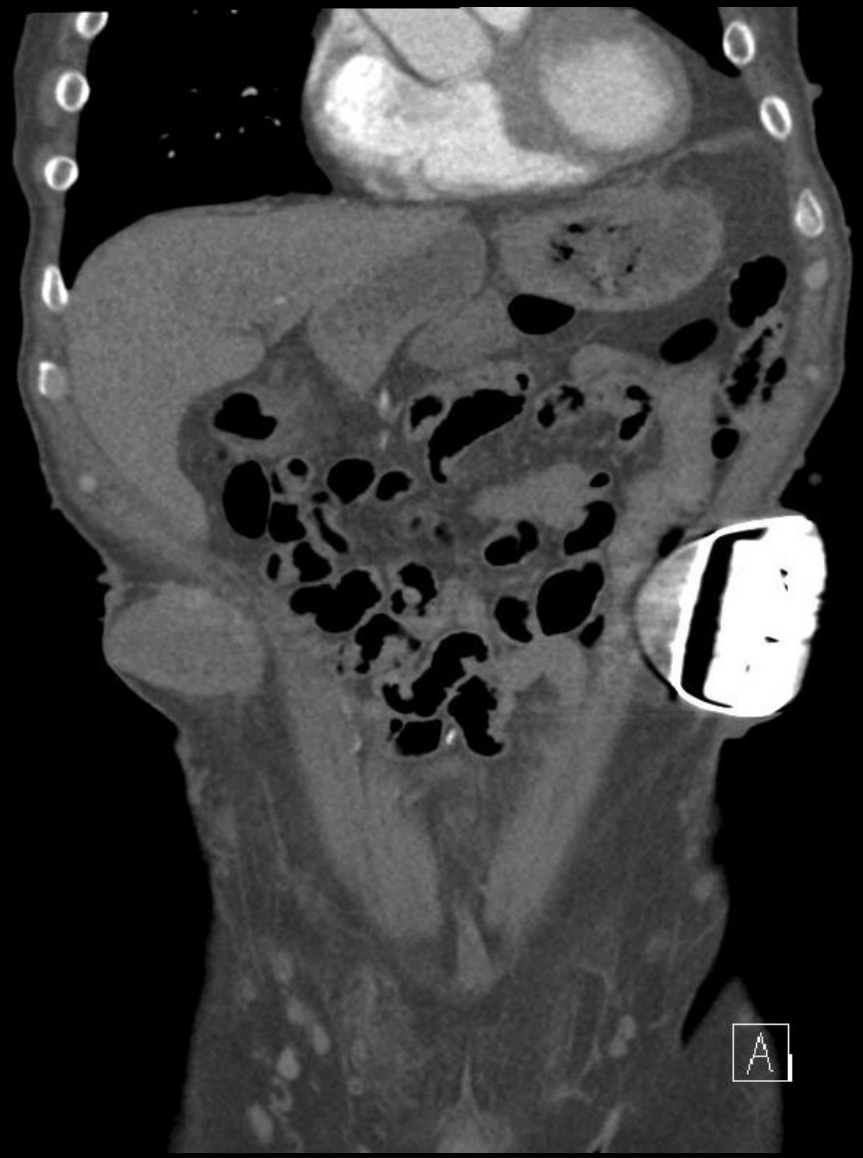
US Abdomen (unlabeled)



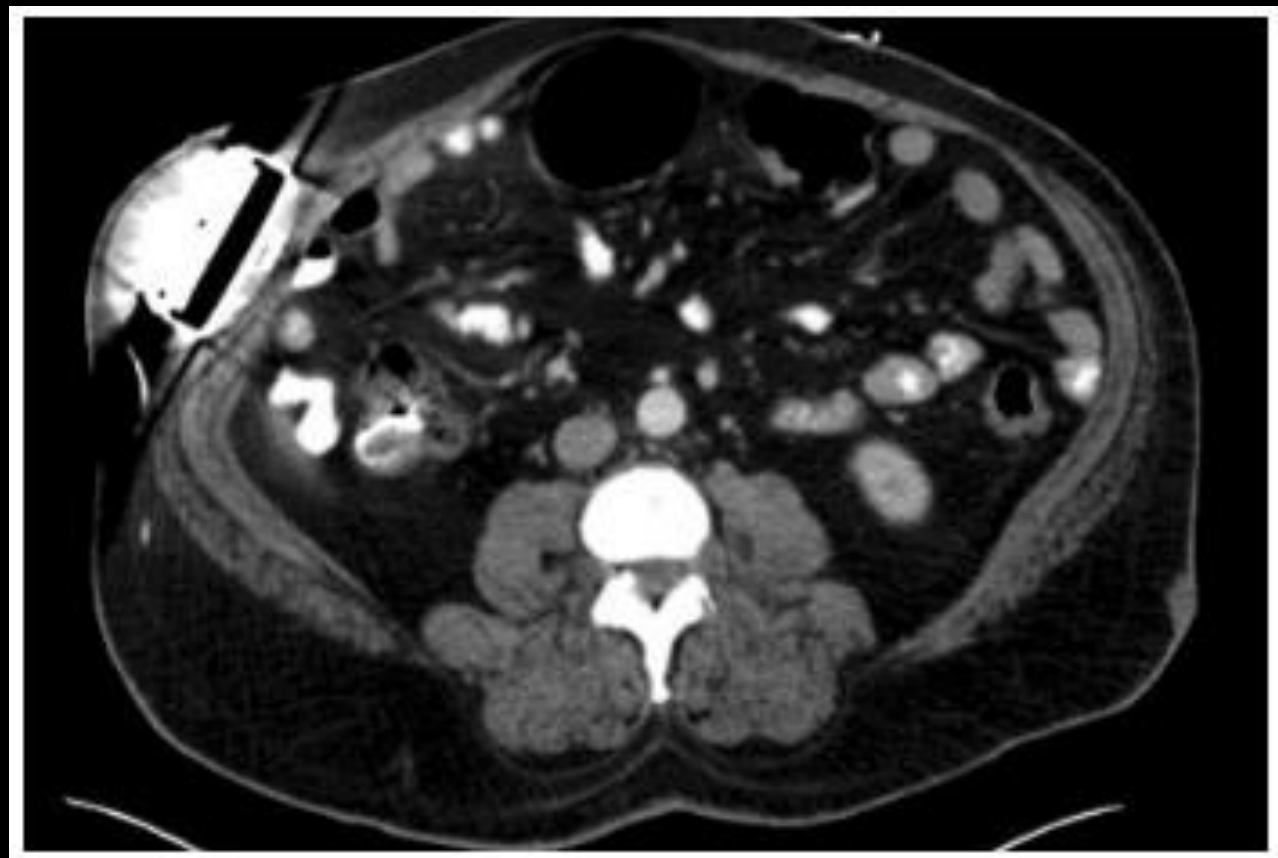
CT Abdomen, with Contrast, Axial (unlabeled)



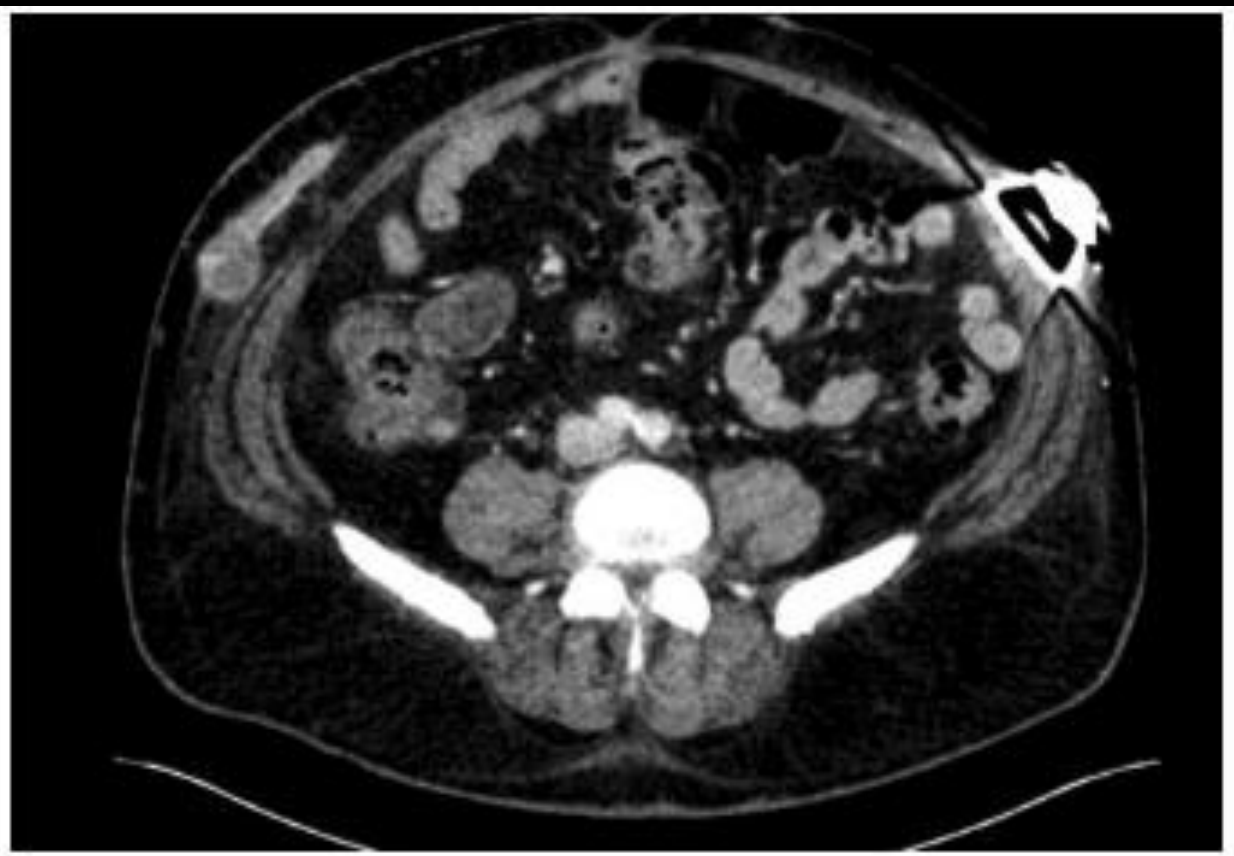
CT Abdomen, with Contrast, Coronal (unlabeled)



Historic CT Abdomen (unlabeled)

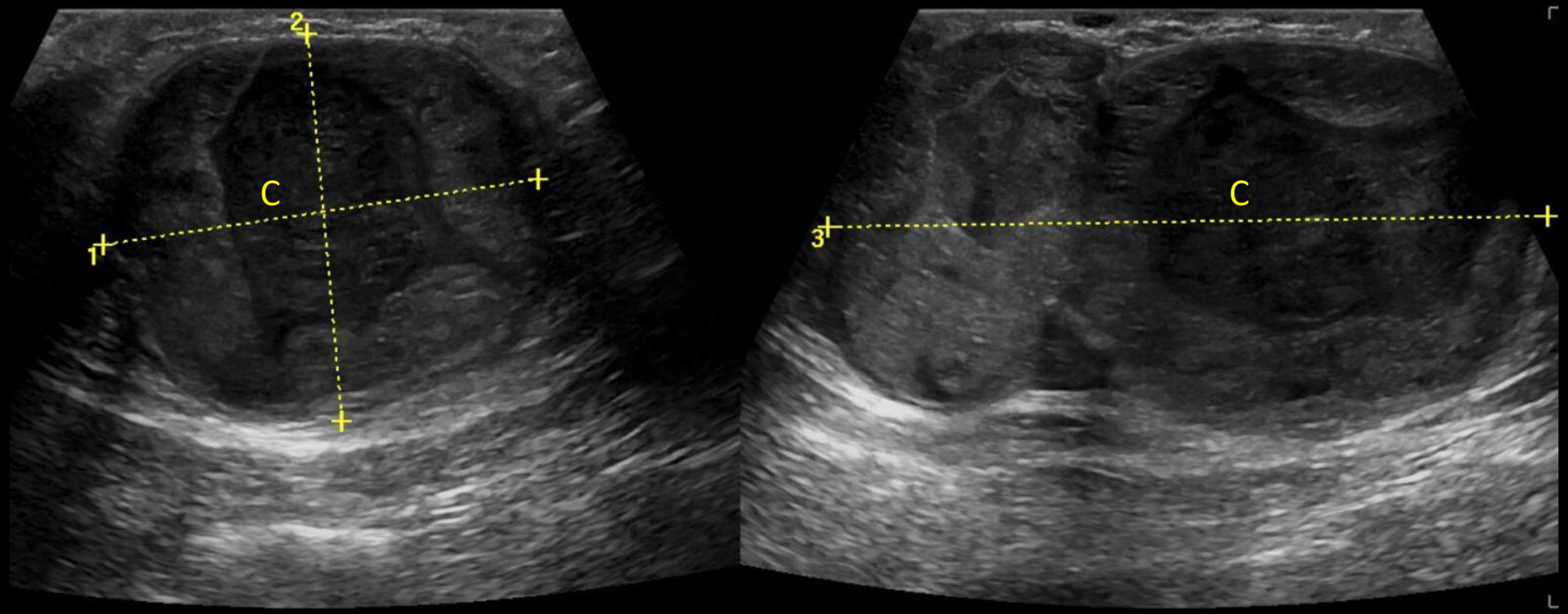


2013



2017

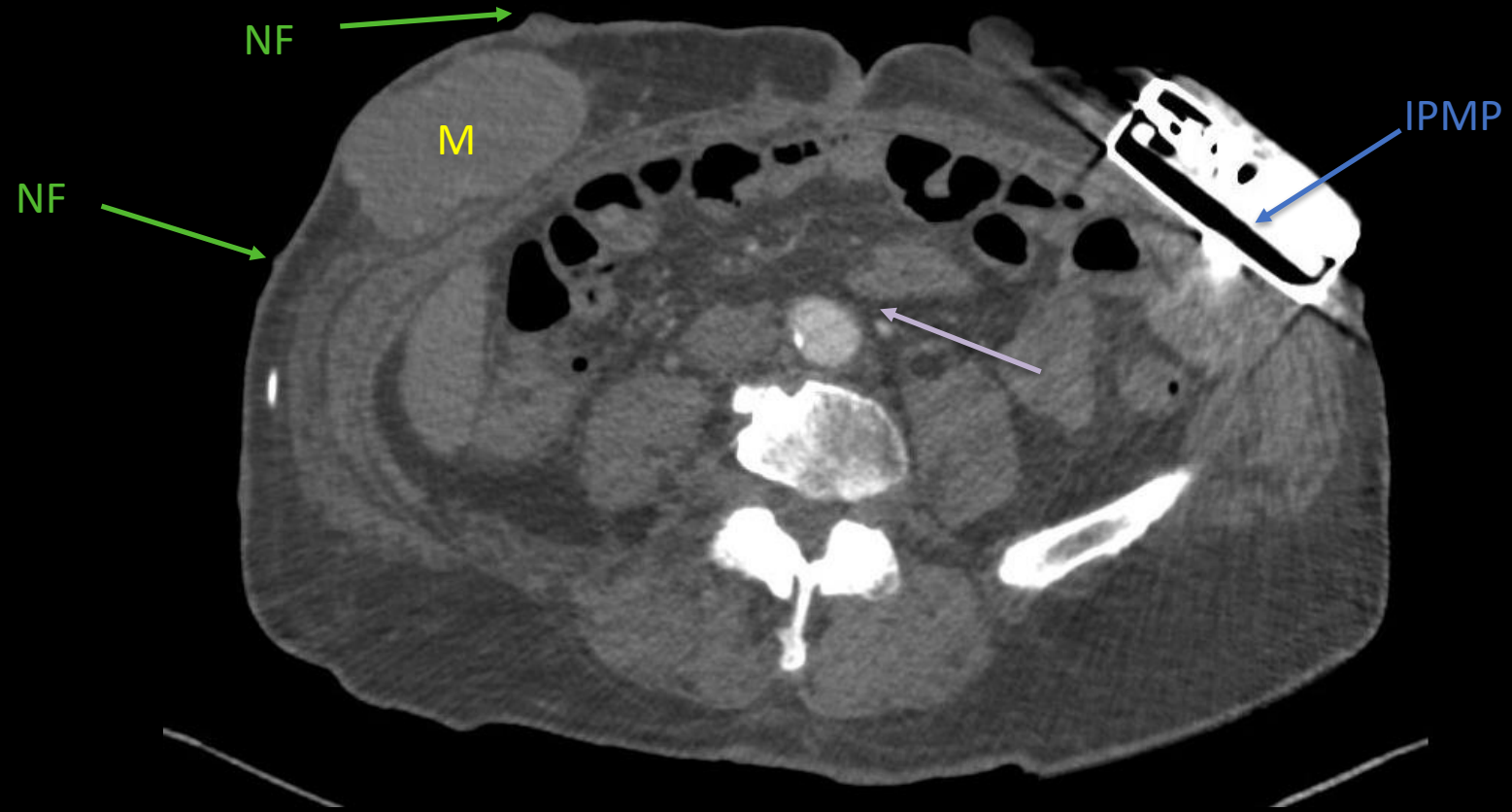
US Abdomen (labeled)



Heterogenous, circumscribed mass measuring 4.4 x 7.2 x 3.9 cm with hypo-echoic center (C).

US appearance concerning for necrosis and malignancy

CT Abdomen with Contrast, Axial (labeled)



Well demarcated enhancing solid mass (M) measuring 7.4 x 3.8 x 4.3 cm localized to the soft tissue of the right flank

Multiple neurofibromas (NF) on the skin

Implanted pain medication pump (IPMP) visible implanted under the skin in the subcutaneous tissue

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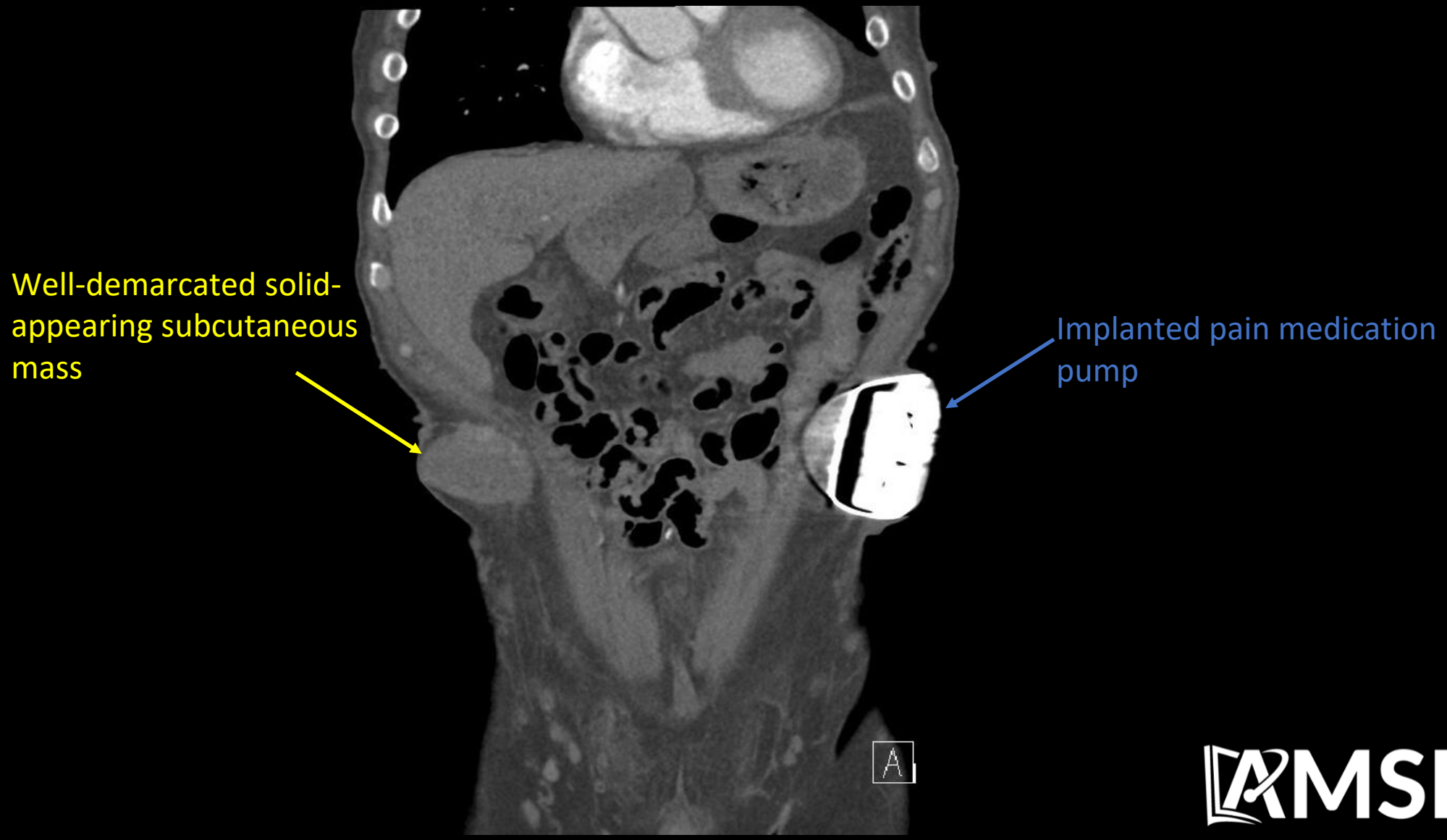
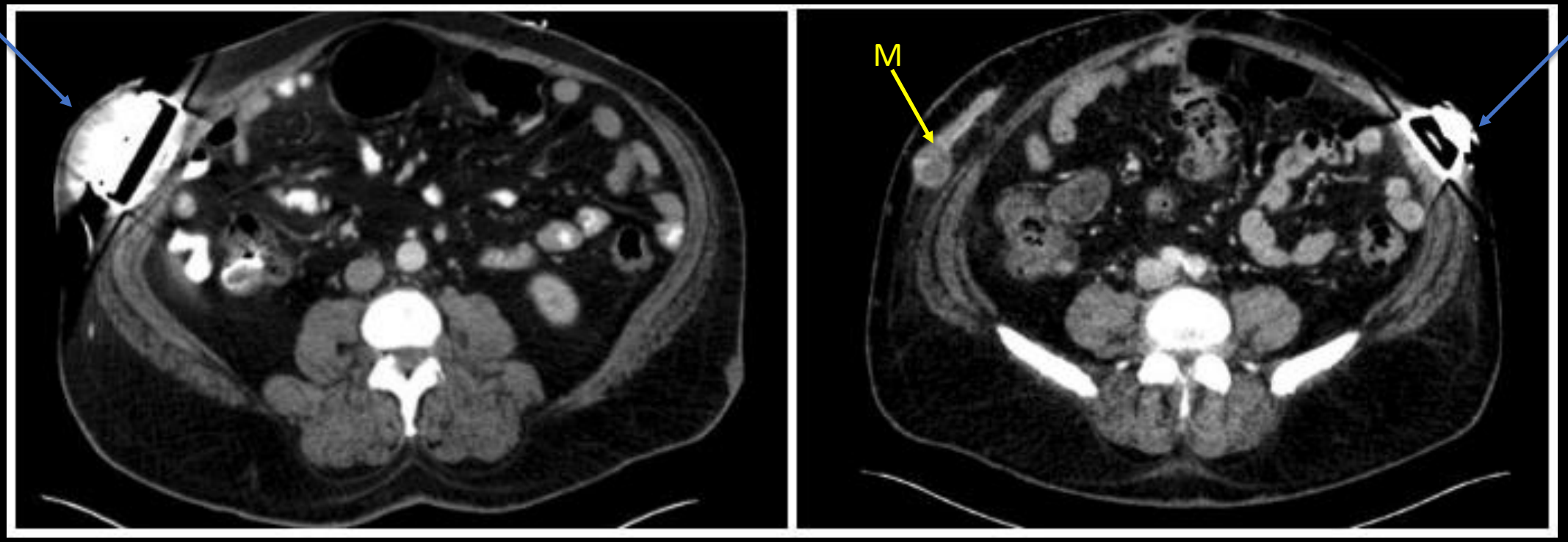
CT Abdomen with Contrast, Coronal (labeled)



Historic CT Abdomen (labeled)



2013

Original implanted pain medication pump (IPMP) can be visualized in the right lower quadrant subcutaneous tissue without associated fluid collection or mass

2017

Second / replacement Implanted pain medication pump placed in the left lower quadrant subcutaneous tissue

Thick walled, peripherally enhancing fluid collection measuring 7.7 x 7.1 x 2.2

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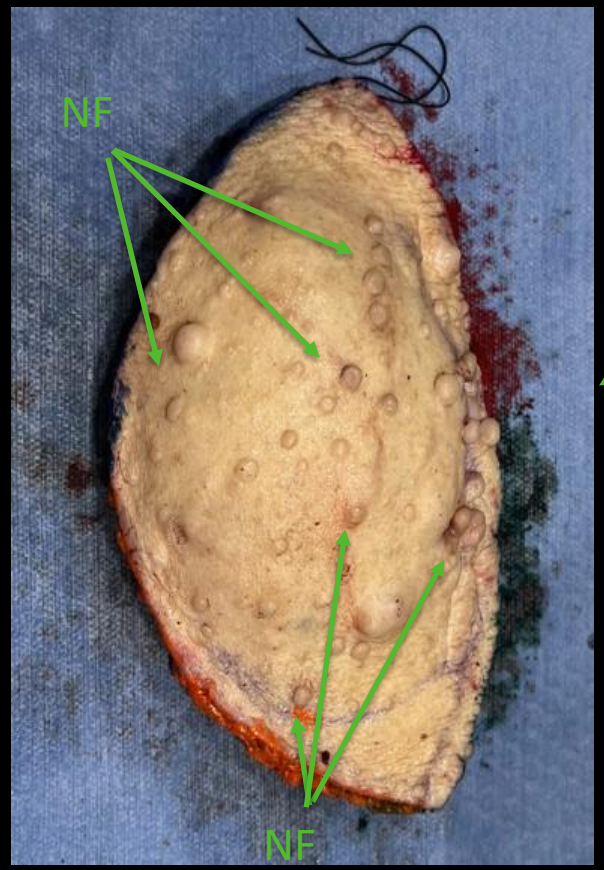
DDX (based on imaging)

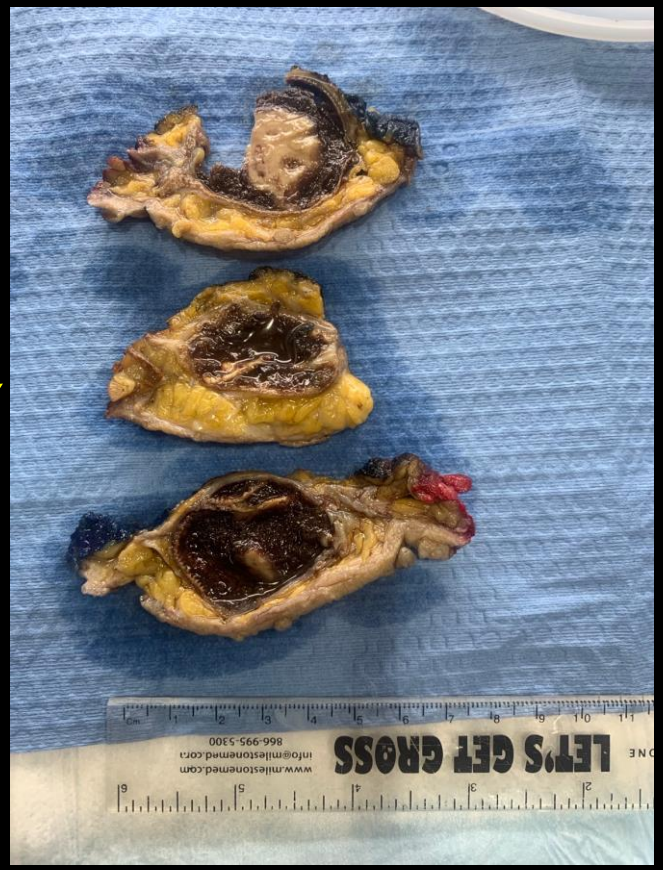
- Cystic mass
- Sarcoma
- Fibromatosis
- Lipoma
- Hamartoma
- Subcutaneous Hematoma

Gross Path (labeled)

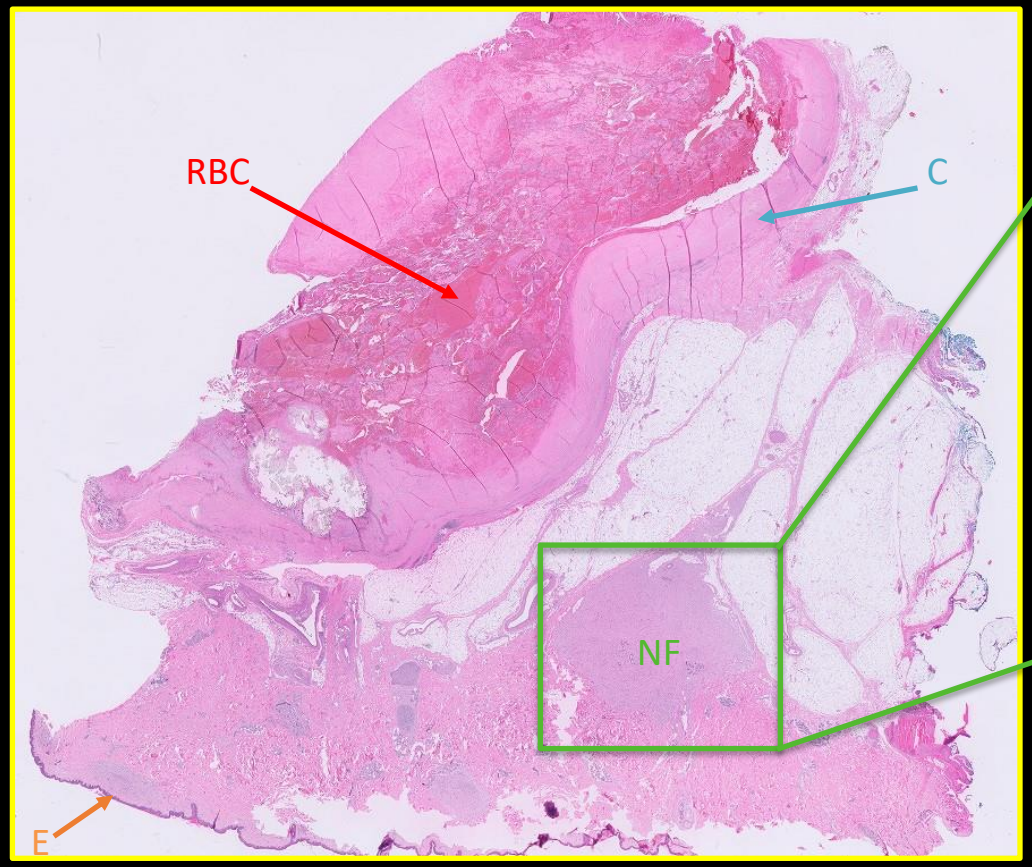


Pedunculated nodular lesions across skin surface of excision specimen, consistent with neurofibromas (NF)

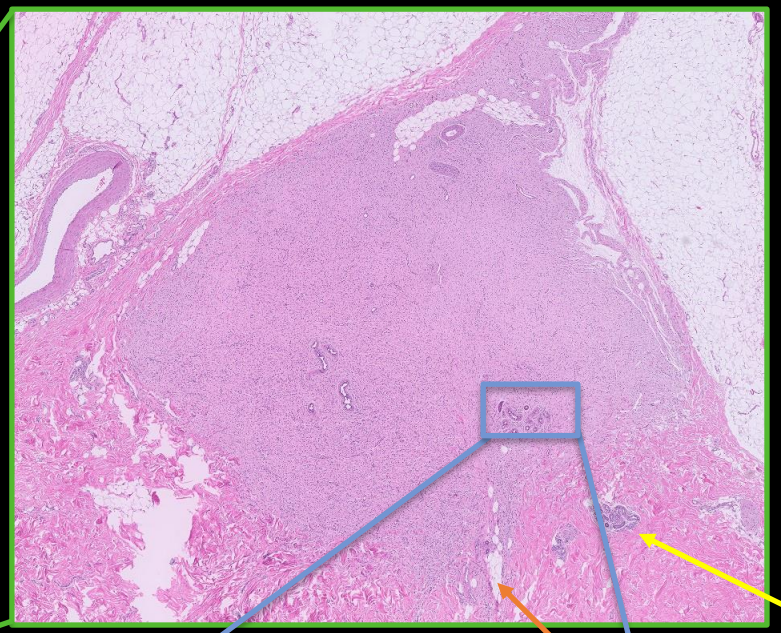
Cut surface of sectioned mass with heterogeneous red-brown to tan-gray appearance



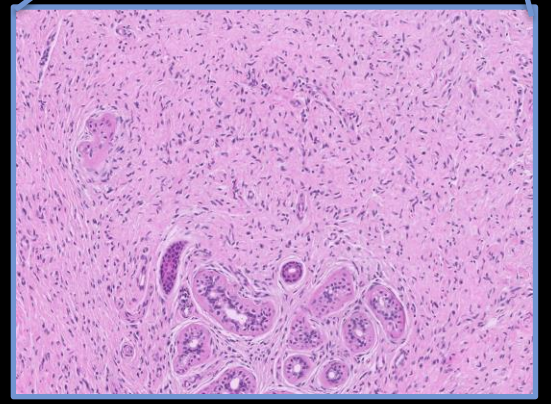
Micro Path (labeled)



H&E section of skin and lesion. Epidermis (E) is visible at the bottom of the image Amphophilic, nodular mass within the dermis consistent with a neurofibroma (NF). A thick, fibrous capsule (C) separates the aforementioned red-brown mass, comprised exclusively of blood products (RBC) in this section, from the subcutaneous fibroadipose tissue.



Higher magnification view of the neurofibroma. This is a cellular lesion entrapping some skin adnexal structures such as sweat glands (G), as well as adipose tissue (A)



Higher power magnification of the neurofibroma. This is a somewhat bland and spindled appearing lesion comprised of an overgrowth of elements of the nerve sheath, mainly Schwann cells and myofibroblasts. Entrapped skin adnexal structures are seen at the bottom of this image, as well.

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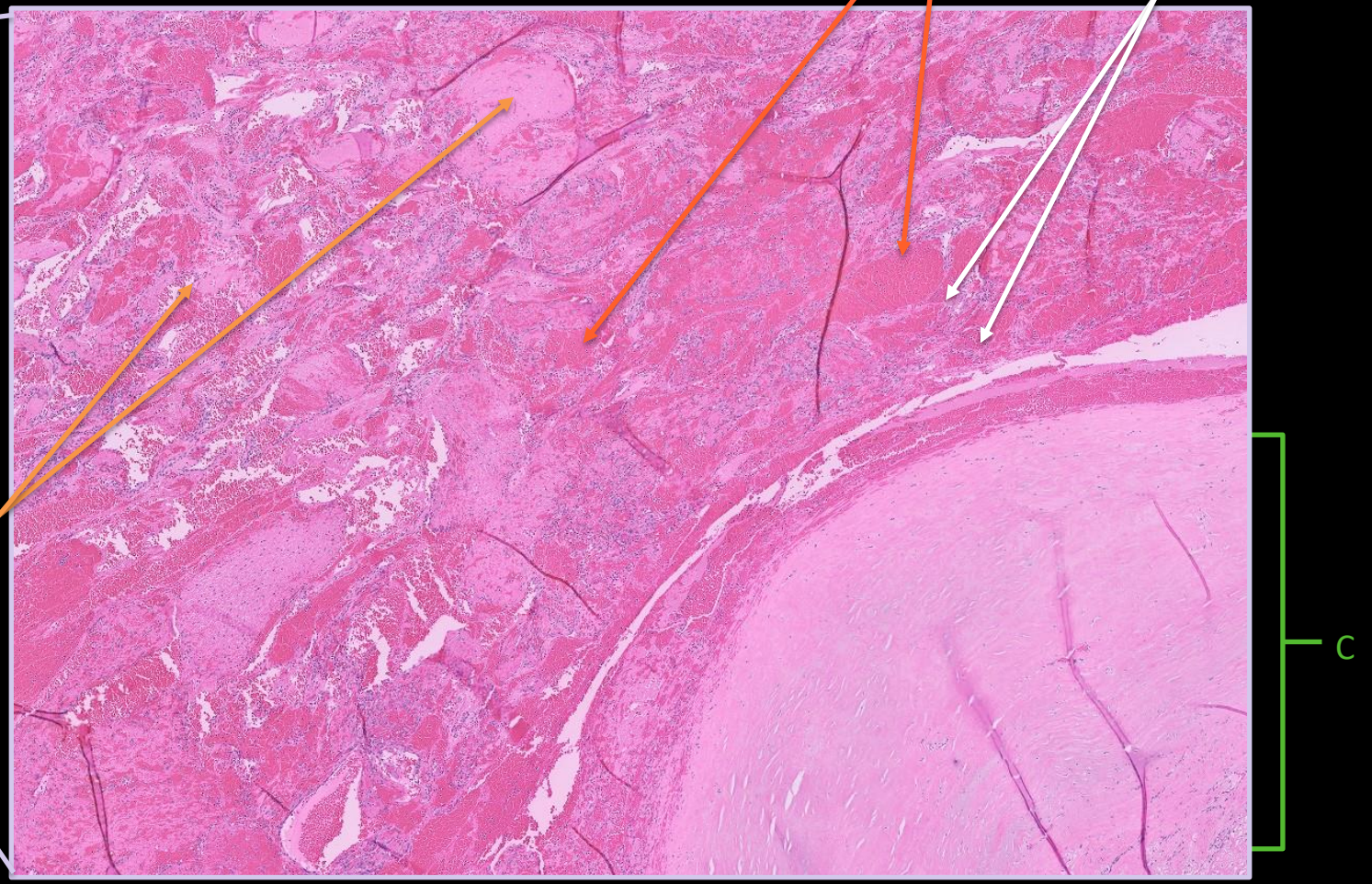
Micro Path (labeled)



Higher magnification of the contents of the red-brown, encapsulated lesion. The lesion is comprised of alternating regions of red blood cells (RBC) and fibrin (F), also known as Lines of Zahn, consistent with a hematoma. In addition there is evidence of organization of the hematoma, as it exhibits neovascularization. The eosinophilic structure at the bottom right is the aforementioned fibrous capsule (C).

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Final Diagnosis:

Resolving Hematoma

Case Discussion:

Neurofibromatosis Type 1

Symptoms & Manifestations

Neurofibromatosis type 1 (NF1) is caused by mutations in the gene NF1, a tumor suppressor gene on chromosome 17 expressed in neurons and glial cells. Manifestations include a constellation of features:

- Cutaneous: Neurofibromas, cafe-au-lait spots, axillary freckling
- Ocular: Lisch nodules, optic gliomas
- MSK: Short stature, scoliosis
- Cardiac: HTN
- Neuro/Psych: ADHD, learning disabilities

Rare accounts of hematomas associated with NF1 have been cited in case reports (typically spontaneous and rapidly enlarging). However, this is not a common finding. This patient's hematoma was slow growing and associated with known insult (prior medication pump and subsequent scar tissue formation)

Inheritance & Screening

- This is a relatively rare condition, affecting 1 in 3000-4000.
- Inheritance is autosomal dominant, and individuals are born with a single gene mutation. However, a second, acquired hit to the affected gene is necessary to cause the physical manifestations, such as neurofibromas.
- Routine screenings for additional manifestations:
  - Ophthalmologic evaluation
  - Blood pressure monitoring
  - Expedited cancer screenings
  - Surgical management for scoliosis if indicated

NF1 & Pain

Chronic pain is the underlying feature leading to this patient's hematoma; i.e. he's had multiple implanted pain medication pumps (IPMP) placed and removed, and has developed subsequent scar tissue and ultimately, a hematoma.

Interestingly, at the time of the surgery, a retained catheter (i.e. foreign body) from the prior IPMP was discovered within the fibrous capsule and removed.

Pain in NF1

- NF1 is highly associated with chronic, diffuse pain, however, the mechanism for this pain is not fully understood.
- Some patients, but not all, associate this pain with their neurofibromas, but others report more widespread pain with no clear source.

Treatment & Further Research

- Some resources report that traditional pain medications, such as opioids, are not effective in treating pain caused by NF1, and pain is often managed with surgery.
- Some researchers are attempting to replace traditional pain management with therapies like Acceptance and Commitment Therapy. mTOR inhibiting agents like sirolimus have also undergone trials for pain treatment with unclear results.

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